Question 1 of 31

Question Difficulty: Easy

12 years old boy was admitted due to acute cardiac failure. He has a history of recurrent pneumothorax.

Examination shows he is a tall boy with an arm span of 1.1 of the body ratio. His trunk shows thoracolumbar scoliosis. His mouth shows high arched palate, with dental crowding. Echo shows aortic root dilatation complicated by aortic valve regurgitation.

What is the most likely diagnosis?

- ○ **Gigantism**
- ○ **Familial tall stature**
- ○ **Homocystinuria**
- ● **Marfan syndrome**
- ○ **Klinefelter syndrome**

Description

Marfan syndrome (MFS) signs and symptoms:

- Facial appearance (dolichocephaly, malar hypoplasia, enophthalmos, retrognathia, down-slanting palpebral fissures)
- Pectus carinatum and excavatum
- Reduced upper-to-lower segment ratio
- Arm span is greater than 1.05
- A positive wrist sign (the thumb and index fingers overlap when encircling the contralateral wrist.)
- A positive thumb (Steinberg) sign (the thumb extends beyond the ulnar border of the hand when the digit is held flexed in the palm)
- A thoracolumbar scoliosis
- Joint hypermobility
- High arched palate, with dental crowding
- Retinal detachment
- Recurrent spontaneous pneumothorax

Cardiovascular complications

- Dilatation of the ascending aorta, with or without aortic valve regurgitation (most common)
- Aortic dissection
- Mitral valve prolapse

Question 2 of 31

Question Difficulty: Average

15 years old boy was admitted due to hematuria and lower limb edema. His mother reported a long history of hearing and visual difficulties.

His eyes show protrusion on the crystalline lens capsule in to the anterior chamber. Fundoscopic examination shows retinopathy.

Investigations show:

Hemoglobin	12 g/dL	(Male: 13.5-17.5 g/dL)
Serum creatinine	186 mmol/L	(53-106 mmol/L)
Serum sodium	129 mmol/L	(136-145 mmol/L)
Serum potassium	4.5 mmol/L	(3.5-5.0 mmol/L)
Serum albumin	28 g/L	(35 - 50 g/L)
24 urine	5.3 gm protein	

Renal biopsy sample shows patches of collapsed glomeruli.

What is the most likely diagnosis?

- ○ **Fabry disease**
- ○ **Alport syndrome**
- ○ **Cystinuria**
- ◉ **Berger syndrome**
- ○ **Bartter Syndrome**

Description

Alport syndrome is an inherited disease (X-linked dominant in 85% of cases) causing defect in the gene which codes for type IV collagen which is an important structural component of basement membranes in the kidney, inner ear and eye. Signs and symptoms include:

- Renal manifestations include hematuria, proteinuria and hypertension (Focal segmental glomerulosclerosis)
- Ocular manifestations include anterior lenticonus (protrusion on the crystalline lens capsule), retinopathy, corneal dystrophy and macular thinning
- Sensori-neural deafness
- Leiomyomatosis (benign smooth muscle tumors) of the esophagus and tracheobronchial tree

Question 3 of 31

Question Difficulty: Average

Mr. X was diagnosed to have a genetic disease. He reported that his father, mother, brothers and sisters are all free of this disease. On questioning his father he reported that his grandfather had a similar condition and his mother reported a similar condition to her uncle.

What is the mode of inheritance of this disease?

- (●) **Autosomal dominant inheritance**
- ( ) **X-linked dominant inheritance**
- ( ) **Mitochondrial inheritance**
- ( ) **X-linked recessive inheritance**
- ( ) **Autosomal recessive inheritance**

Description

Autosomal recessive inheritance features:

- Males and females are equally affected, but are fewer in number than in autosomal dominant conditions.
- Not all generations will be affected.
- If both parents are carriers for the recessive gene 25% of the offspring will be affected, 50% will become carriers, but will not have the disease and 25% will not have the abnormal gene.
- If an affected individual marries a carrier, then 50% off -spring will be affected and 50% offspring will be carriers.
- If an affected individual marries another affected, then all offspring will be affected

CHECK ANSWER END SESSION NEXT

Session Statistics

Answered: 2 ✓ Correct: 1 × Incorrect: 1 Average Score: 50 %

Question 4 of 31

Question Difficulty: Hard

Mr. X is seeking medical advice. He mentioned that his father and his mother had an inherited disease and his mother and her family were free of it. He reported that he is free of the disease and he has a free sister and affected sister and brother.

What is the mode of inheritance of this disease?

- Mitochondrial inheritance
- X-linked dominant inheritance
- Autosomal recessive inheritance
- X-linked recessive inheritance
- Autosomal dominant inheritance

Description

Autosomal dominant inheritance features:

- Males and females are equally affected.
- All inheriting the abnormal gene are affected.
- Offspring of affected parents (irrespective of the parental sex) have a 50% chance of inheriting the disease.

Autosomal recessive inheritance should be excluded as the affected father and free mother with no family history will have no chance in having affected children.

CHECK ANSWER END SESSION NEXT

Session Statistics

Answered: 3 ✓ Correct: 1 × Incorrect: 2 Average Score: 33.33 %

Question 5 of 31

Question Difficulty: Hard

Mrs. X is complaining of severe abdominal pain, nausea and vomiting. She has a long history of epilepsy. She reported darkening of urine on standing.

Examination shows proximal muscle weakness. Investigations show hyponatremia and elevated WBCs.

What is the management of choice?

- Insulin
- Domperidone
- Chloroquine
- Intravenous fluids
- Hematin

Description

This patient is complaining of acute intermittent porphyria. It is an autosomal dominant condition caused by defect in porphobilinogen deaminase. It is typically presented by abdomen pain, neuropathies (paralysis and seizures) and constipation, but without skin rash.

Complications include hyponatremia due to SIADH and mild leukocytosis.

High doses of glucose can inhibit heme synthesis and are useful for treatment of mild attacks. IV hematin (Heme arginate) is of choice for severe attacks and neurologic symptoms.

CHECK ANSWER END SESSION NEXT

Session Statistics

Answered: 4 ✓ Correct: 1 ✕ Incorrect: 3 Average Score: 25 %

Please wait until the operation has finished (Hit Esc to cancel)

Question 6 of 31

Question Difficulty: Hard

16 years old girl is suffering from primary amenorrhea. Examination showed she has short stature. There is total absence of breast development and secondary sexual characters.

There is excessive nevi and webbed neck. Her hand and feet show pitting edema and there are short fourth and fifth metacarpals and metatarsals.

What is the most likely diagnosis?

- (●) Pseudoxanthoma elasticum
- ( ) Osteogenesis imperfecta
- ( ) Marfan syndrome
- ( ) Noonan syndrome
- ( ) Turner syndrome

Description

Turner syndrome is caused by the absence of one set of genes from the short arm of one X chromosome. Signs and symptoms include:

- Short stature

Question 6 of 31

Question Difficulty: Hard

16 years old girl is suffering from primary amenorrhea. Examination showed she has short stature. There is total absence of breast development and secondary sexual characters.

There is excessive nevi and webbed neck. Her hand and feet show pitting edema and there are short fourth and fifth metacarpals and metatarsals.

What is the most likely diagnosis?

- (●) **Pseudoxanthoma elasticum**
- ( ) **Osteogenesis imperfecta**
- ( ) **Marfan syndrome**
- ( ) **Noonan syndrome**
- ( ) **Turner syndrome**

Description

Turner syndrome is caused by the absence of one set of genes from the short arm of one X chromosome. Signs and symptoms include:

- Short stature
- Absent breast development
- Primary ovarian failure
- Excessive Nevi
- Webbed neck
- Scoliosis
- Swollen hands and feet because of lymphedema
- High arched palate with dental crowding or malocclusion
- Short fourth and fifth metacarpals and metatarsals
- Coarctation of the aorta (most common cardiac complication), bicuspid aortic valve and aortic dissection
- Hypothyroidism and goiter

CHECK ANSWER END SESSION NEXT

Question 7 of 31

Question Difficulty: Easy

12 years old child is complaining of a hemolytic disease and it was suspected it is a genetic disease. Genetic analysis of the family showed an affected brother and free brother and sister. He has an affected uncle from the mother side and his father and his family are free of the disease.

What is the mode of inheritance of this disease?

- X-linked recessive inheritance
- Autosomal dominant inheritance
- Mitochondrial inheritance
- Autosomal recessive inheritance
- X-linked dominant inheritance

Description

Tis boy has G6PD disease. It is an X-linked recessive inheritance disease. Features include:

- Only males are affected but not carriers
- No male-to-male transmission
- Females are carriers
- Daughters of affected males will be carriers
- Female carrier will have 50% affected sons and 50% daughters who are carriers

CHECK ANSWER END SESSION NEXT

Session Statistics

Answered: 6 ✓ Correct: 1 ✕ Incorrect: 5 Average Score: 16.67 %

Question 8 of 31

Question Difficulty: Average

Mr. X is seeking medical advice. He mentioned that his father and his mother had an inherited disease and his mother and her family were free of it. He reported that he is free of the disease and he has a free sister and affected sister and brother.

What is the chance for his children in having the disease if his wife and her family are clear of the disease?

- 100%
- 50%
- 75%
- 0%
- 25%

Description

This disease is an autosomal dominant disease. Mr. X is clear of the disease and is not a carrier as his wife; so all their children will be free of this disease.

Autosomal recessive inheritance should be excluded as the affected father and free mother with no family history will have no chance in having affected children.

CHECK ANSWER END SESSION NEXT

Session Statistics

Answered: 7 ✓ Correct: 1 × Incorrect: 6 Average Score: 14.29 %

Question 9 of 31

Question Difficulty: Hard

4 month old infant was admitted due to seizure for the first time. Examination shows normally developed infant with slight hypopigmented hair and skin. He has a musty odor.

Which of the followings could be diagnostic if found in urine?

- Arginine
- Homocysteine
- Ornithine
- Phenyl pyruvic acid
- Cysteine

Description

Phenylketonuria (PKU) is an autosomal recessive condition caused due to defect in phenylalanine hydroxylase, an enzyme located on chromosome 12 which converts phenylalanine to tyrosine. Signs and symptoms include:

- Infants with PKU are normal at birth
- Seizures
- Hypopigmentation (excessively fair hair and skin)
- Musty odor of the baby's sweat and urine
- Permanent brain damage (learning difficulties and intellectual disability) occurs if PKU is untreated during the first months of life

Phenyl pyruvic acid in urine is diagnostic.

CHECK ANSWER END SESSION NEXT

Session Statistics

Answered: 8 ✓ Correct: 1 ✗ Incorrect: 7 Average Score: 12.5 %

Question 10 of 31

Question Difficulty: Hard

25 years Mr. X is complaining of visual disturbances. Genetic analysis revealed that his mother and all his brothers and sisters have similar condition. His father`s family are clear of the disease.

What is the chance of transferring the disease to his offspring?

- 100% chance of all offspring
- 50% chance of all offspring
- Males will be carriers and females will have the disease
- 0% chance of all offspring
- Females will be carriers and males will have the disease

Description

This is a mitochondrial genetic abnormality due to Leber optic atrophy. Features of mitochondrial genetic abnormalities include:

- During conception, only mitochondria from the ovum are passed on to the zygote.
- Affected females will pass the disease to 100% off spring
- Affected males will not transmit disease to offspring.

CHECK ANSWER END SESSION NEXT

Session Statistics

Answered: 9 ✓ Correct: 2 × Incorrect: 7 Average Score: 22.22 %

Question 11 of 31

Question Difficulty: Hard

Mr. X was diagnosed to have renal cell carcinoma. He reported his brother was diagnosed with the same condition 3 years ago and his father died due to cerebral stroke.

What is the most likely diagnosis?

- (●) **Gaucher disease**
- ( ) **Thrombotic thrombocytopenic purpura**
- ( ) **Polycystic kidney disease**
- ( ) **Von Hippel–Lindau disease**
- ( ) **Alport syndrome**

Description

Von Hippel-Lindau (VHL) is an autosomal dominant disease due to an abnormality in the VHL gene located on short arm of chromosome 3 causing visceral cysts and benign tumors in multiple organ systems. Signs and symptoms include:

- CNS (cerebellar, spinal) hemangioma
- Retinal hemangioma causing vitreous hemorrhage
- Renal cysts with increased risk for transformation into renal cell carcinoma
- Pheochromocytoma
- Pancreatic cysts

Polycystic kidney disease is not complicated by renal cell carcinoma.

CHECK ANSWER END SESSION NEXT

Session Statistics

Answered: 10 ✓ Correct: 2 ✗ Incorrect: 8 Average Score: 20 %

Question 12 of 31

Question Difficulty: Easy

Mr. X is complaining of exertional angina which has deteriorated over the last year. He also tingling sensation in his hands and feet. He has a long history of hypertension.

Examination shows lower limb edema and finger nails show Raynaud disease. Neurological examination shows glove and stocking sensory loss. His urine dipstick shows hematuria and ++ proteinuria.

What is the most likely diagnosis?

- (●) **Mitochondrial disorder**
- ( ) **Fabry disease**
- ( ) **Tuberose sclerosis**
- ( ) **Homocysteinuria**
- ( ) **Autosomal dominant polycystic kidney disease**

Description

Fabry disease is an X-linked lysosomal disorder that leads to excessive deposition of neutral glycosphingolipids in the vascular endothelium of several organs and in epithelial and smooth muscle cells. Signs and symptoms include:

- Acroparesthesia (Full body or localized pain to the extremities)
- Progressive renal failure
- Hypertension and cardiomyopathy
- Angiokeratomas (tiny, painless papules)
- Anhidrosis
- Raynaud disease
- Peripheral neuropathy

CHECK ANSWER END SESSION NEXT

Session Statistics

Question 13 of 31

Question Difficulty: Hard

Mr. X is complaining of rectal bleeding and a change in bowel habit. Je reported that his father died due to a colonic carcinoma. He has a previous history of osteomas of both tibia and skull.

Examination shows multiple skin fibromas. Colonoscopy was done and showed multiple benign adenomatous polyps in the colon.

What is the most likely diagnosis?

- Kartagener syndrome
- Turcot syndrome
- Peutz-Jegher syndrome
- Familial adenomatous polyposis
- Gardener syndrome

Description

Gardner syndrome is an autosomal dominant subtype of familial adenomatous polyposis. It us also manifested by extracolonic tumors as osteomas, thyroid cancer, epidermoid cysts and fibromas.

CHECK ANSWER END SESSION NEXT

Session Statistics

Answered: 12 ✓ Correct: 2 ✕ Incorrect: 10 Average Score: 16.67 %

Question 14 of 31

Question Difficulty: Average

6 years ol child was admitted due to dislocation of his right shoulder and right hip after a minor fall. He has a long history of recurrent joint dislocations

Examination shows he has short stature. His skin was very lax with multiple bruising.

What is the most likely diagnosis?

- Acromegaly
- Homocystinuria
- Ehlers-Danlos syndrome
- McCune-Albright syndrome
- Marfan syndrome

Description

Ehlers-Danlos syndrome is a congenital disorder causing collagen synthesis abnormalities. Signs and symptoms include:

- Generalized joints hypermobility and dislocations
- Skin laxity
- Easy bruising and ecchymosis
- Scoliosis
- Short stature
- Aortic dissection

CHECK ANSWER END SESSION NEXT

Session Statistics

Answered: 13 ✓ Correct: 2 × Incorrect: 11 Average Score: 15.38

Question 15 of 31

Question Difficulty: Hard

Mr. X is complaining of multiple lesions over his arm and trunk that is shown in the image. He also has several darker brown large macules on her trunk. There are tiny freckle-like lesions in both axillae.

As a part of diagnosis; which of the following would be found?

- Pheochromocytoma
- Lisch nodules
- Astrocytoma
- Charcot joints
- Acoustic neuroma

As a part of diagnosis; which of the following would be found?

- Pheochromocytoma
- Lisch nodules
- Astrocytoma
- Charcot joints
- Acoustic neuroma

Description

This patient has Neurofibromatosis type I (Recklinghausen syndrome). It is caused by a gene mutation on chromosome 17 which encodes neurofibromin. Clinical diagnosis requires the presence of at least 2 of 7 criteria:

- Six or more café-au-lait spots
- Two or more Axillary or inguinal freckles
- Two or more typical neurofibromas (shown in the image)
- Two or more Lisch nodules (iris hamartoma)
- Optic nerve glioma
- Sphenoid dysplasia or typical long-bone abnormalities such as pseudarthrosis
- First-degree relative with NF1

CHECK ANSWER END SESSION NEXT

Session Statistics

Answered: 14 ✓ Correct: 2 ✕ Incorrect: 12 Average Score: 14.29 %

Question 16 of 31

Question Difficulty: Easy

Mr. X was admitted due to severe hematemesis. He mentioned he had previous colonoscopy for diagnosis of bleeding per rectum and previous admission due to He also reported his mother died due to cerebral stroke.

Examination shows multiple spider veins on his face and neck.

What is the most likely diagnosis?

- Peutz-Jegher syndrome
- Hereditary hemorrhagic telangiectasia
- Cerebral amyloid angiopathy
- Wegener granulomatosis
- Intracranial aneurysm

Description

Hereditary hemorrhagic telangiectasia (Osler-Weber-Rendu disease) is an autosomal dominant genetic disorder that leads to abnormal blood vessel formation in the skin, mucous membranes and organs.

The 4 clinical diagnostic criteria are:

- Family history (a first-degree relative with HHT)
- Epistaxis
- Telangiectasias in the skin and mucosal linings of the nose and gastrointestinal tract
- Arteriovenous malformations occur in larger organs as the lungs (hemoptysis), liver and the brain (strokes)

CHECK ANSWER END SESSION NEXT

Session Statistics

Answered: 15 ✓ Correct: 2 × Incorrect: 13 Average Score: 13.33

Question 17 of 31

Question Difficulty: Hard

Mr. X is admitted due to his left flank that radiates down to his groin with associated dysuria and offensive urine with the odor of rotten eggs.

Abdominal ultrasound confirmed a left ureteric stone. He reported lifelong recurrent stones and all previous management measures fail.

What is the management of choice?

- (●) Furosemide
- ( ) Bendrofluazide
- ( ) Penicillamine
- ( ) Desferrioxamine
- ( ) Allopurinol

Description

This patient is complaining of Cystinuria. It is an autosomal-recessive defect in reabsorption of cysteine and the dibasic amino acids ornithine, arginine, and lysine from the luminal fluid of the renal proximal tubule and small intestine.

It is characterized by lifelong, recurrent urolithiasis that is difficult to manage, either surgically or medically. The urine has the characteristic odor of rotten eggs.

Management is by large-volume fluid intake, urinary alkalization with potassium citrate and chelation by penicillamine.

CHECK ANSWER END SESSION NEXT

Session Statistics

Answered: 16 ✓ Correct: 2 × Incorrect: 14 Average Score: 12.5 %

Question 18 of 31

Question Difficulty: Easy

12 years old boy is complaining of progressive weakness of both legs over the last 3 years. He has a histoy of auditory and visual difficulties as well.

Neurological examination revealed a loss of pinprick sensation to the knee bilaterally, and weakness of both legs. There is ataxia in lower limbs. Eye examination shows cataract and nystagmus.

Which of the following investigations would be diagnostic?

- (●) **Phytanic acid level**
- ( ) **Genetic testing**
- ( ) **ANCA**
- ( ) **Vitamin E level**
- ( ) **Thyroid function tests**

Description

Refsum disease is an autosomal recessive neuro-cutaneous disease that results from the over-accumulation of phytanic acid in cells and tissues. Signs and symptoms include:

- Ichthyosis (dry, thickened, scaly skin)
- Cardiomyopathy

Neurological symptoms

- Intermittent sensorimotor polyneuropathy
- Anosmia
- Sensorineural deafness
- Cerebellar ataxia

Eye symptoms

- Cataract
- Nystagmus
- Retinitis pigmentosa causing concentric constriction of the visual fields and night blindness

CHECK ANSWER END SESSION NEXT

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Question 19 of 31

Question Difficulty: Easy

12 years old boy is suffering from sudden deterioration of vision in his left eye. He has learning difficulties and past history of deep vein thrombosis.

Examination shows marfanoid appearance with tall and slim body. He has blonde sparse and brittle hair. Slit lamp examination shows downward dislocation of lens.

What is the most likely diagnosis?

- Cystinuria
- Moya Moya disease
- Homocystinuria
- Marfan syndrome
- Fibromuscular dysplasia

Description

Homocystinuria is an autosomal recessively inherited defect in cystathionine synthetase of methionine metabolism, leading to an abnormal accumulation of homocysteine and its metabolites (homocystine, homocysteine-cysteine complex, and others) in blood and urine.

Signs and symptoms include:

- Downward dislocation of lens
- Marfanoid habitus
- Mental retardation
- Thromboembolic complications
- Pes excavatum, pes carinatum, and genu valgum
- CNS strokes
- Blonde sparse and brittle hair

CHECK ANSWER END SESSION NEXT

Question 20 of 31

Question Difficulty: Average

16 years old girl is suffering from primary amenorrhea. Examination showed she has short stature. There is total absence of breast development and secondary sexual characters.

There is excessive nevi and webbed neck. Her hand and feet show pitting edema and there are short fourth and fifth metacarpals and metatarsals. Karyotyping shows 45,X0.

What is the management of choice?

- Dianette
- Pulsatile GnRH
- Life-long LH
- Life-long FSH
- Life-long estrogen

Description

This girl is complaining of Turner syndrome. Turner syndrome is caused by the absence of one set of genes from the short arm of one X chromosome.

Life-long estrogen is indicated for development of female secondary sex characters. Infertility is permanent.

CHECK ANSWER END SESSION NEXT

Session Statistics

Answered: 19 ✓ Correct: 3 × Incorrect: 16 Average Score: 15.79 %

Question 21 of 31

Question Difficulty: Easy

Mr. X is suffering from sudden loss of vision in his right eye. He has a history of cerebellar hemorrhage due to cerebellar cyst and hematuria due to renal cysts.

Fundoscopic examination shows bilateral retinal angiomas with partial retinal detachment in his right eye.

What is the most likely diagnosis?

- (●) **Von Hippel-Lindau disease**
- ( ) **Simple traumatic retinal detachment**
- ( ) **Polycystic kidney disease**
- ( ) **Alport syndrome**
- ( ) **McCune-Albright syndrome**

Description

Von Hippel-Lindau (VHL) is an autosomal dominant disease due to an abnormality in the VHL gene located on short arm of chromosome 3 causing visceral cysts and benign tumors in multiple organ systems. Signs and symptoms include:

- CNS (cerebellar, spinal) hemangioma
- Retinal hemangioma causing vitreous hemorrhage
- Renal cysts with increased risk for transformation into renal cell carcinoma
- Pheochromocytoma
- Pancreatic cysts

CHECK ANSWER END SESSION NEXT

Session Statistics

Answered: 20 ✓ Correct: 3 × Incorrect: 17 Average Score: 15 %

Question 22 of 31

Question Difficulty: Hard

15 years old boy was admitted due to hematuria and lower limb edema. His mother reported a long history of hearing and visual difficulties.

His eyes show protrusion on the crystalline lens capsule in to the anterior chamber. Fundoscopic examination shows retinopathy.

Investigations show:

Hemoglobin	12 g/dL	(Male: 13.5-17.5 g/dL)
Serum creatinine	186 mmol/L	(53-106 mmol/L)
Serum sodium	129 mmol/L	(136-145 mmol/L)
Serum potassium	4.5 mmol/L	(3.5-5.0 mmol/L)
Serum albumin	28 g/L	(35 - 50 g/L)
24 urine	5.3 gm protein	

What would appear in a renal biopsy?

- ○ **Amyloidosis**
- ◉ **Focal segmental glomerulosclerosis**
- ○ **Rapidly progressive glomerulonephritis**
- ○ **Membranous glomerulonephritis**
- ○ **Mesangiocapillary glomerulonephritis**

Description

Alport syndrome signs and symptoms include:

- Renal manifestations include hematuria, proteinuria and hypertension (Focal segmental glomerulosclerosis)
- Ocular manifestations include anterior lenticonus (protrusion on the crystalline lens capsule), retinopathy, corneal dystrophy and macular thinning
- Sensori-neural deafness
- Leiomyomatosis (benign smooth muscle tumors) of the esophagus and tracheobronchial tree

CHECK ANSWER END SESSION NEXT

Question 23 of 31

Question Difficulty: Hard

Mr. X is seeking medical advice about a genetic disease affected his son. He and the mother reported that it is the first time a similar disease to appear in the family from the father and the mother side.

Which of the following statements is correct?

- The mother is a carrier of an autosomal dominant disease
- The mother is a mitochondrial disease carrier
- The son has an X-linked recessive disease
- The father is a carrier of an autosomal recessive disease
- The son has an X-linked dominant disease

Description

Autosomal recessive inheritance features:

- Males and females are equally affected, but are fewer in number than in autosomal dominant conditions.
- Not all generations will be affected.
- If both parents are carriers for the recessive gene 25% of the offspring will be affected, 50% will become carriers, but will not have the disease and 25% will not have the abnormal gene.
- If an affected individual marries a carrier, then 50% off -spring will be affected and 50% offspring will be carriers.
- If an affected individual marries another affected, then all offspring will be affected

CHECK ANSWER END SESSION NEXT

Session Statistics

Answered: 22 ✓ Correct: 5 × Incorrect: 17 Average Score: 22.73 %

Question 24 of 31

Question Difficulty: Easy

Mr. X with a history of epileptic fit was seeking medical advice about brown macules that appeared over his neck and trunk.

Examination also shows he has three patches of hypopigmented skin, acne-like eruption on his face and his trunk showed this patch.

What is the most likely diagnosis?

- (●) Osler disease
- ( ) Multiple endocrine neoplasia type 1
- ( ) Tuberous sclerosis
- ( ) Neurofibromatosis
- ( ) Refsum disease

What is the most likely diagnosis?

- (●) Osler disease
- ( ) Multiple endocrine neoplasia type 1
- ( ) Tuberous sclerosis
- ( ) Neurofibromatosis
- ( ) Refsum disease

Description

Cutaneous complications of Tuberous sclerosis include:

- Adenoma sebaceum (acne-like eruption)
- Depigmented ash-leaf spots
- Shagreen patches (roughened patches over lumbar spine)
- Cafe-au-lait spots

Question 25 of 31

Question Difficulty: Hard

Mr. X is complaining of light brown discolorations on his skin. Examination shows he has multiple light brown pigmented areas on her skin, a number of friable cutaneous skin lesions and axillary freckling.

Slit lamb examination was shown in the image

What is the most likely diagnosis?

- Neurofibromatosis Type 1
- Neurofibromatosis Type 2
- Acanthosis nigricans
- MEN-2
- MEN-1

Description

Neurofibromatosis is an autosomal dominant disorder that disturbs cell growth in your nervous system, causing tumors to form on nerve tissue.

Neurofibromatosis type I (Recklinghausen syndrome) is caused by a gene mutation on chromosome 17 which encodes neurofibromin. Clinical diagnosis requires the presence of at least 2 of 7 criteria:

- Six or more café-au-lait spots
- Two or more Axillary or inguinal freckles
- Two or more typical neurofibromas
- Two or more Lisch nodules (iris hamartoma) shown in the image
- Optic nerve glioma
- Sphenoid dysplasia or typical long-bone abnormalities such as pseudarthrosis
- First-degree relative with NF1

Neurofibromatosis type II is caused by gene mutation on chromosome 22; it causes bilateral acoustic neuroma and café-au-lait spots.

Question 26 of 31

Question Difficulty: Easy

18 years old girl was referred due to hypertension. She has a history of primary amenorrhea and short stature.

Examination shows absent breast development and secondary sexual characters. There is excessive nevi and webbed neck.

What is the most likely cause for her hypertension?

- Renal artery stenosis
- **Coarctation of the aorta**
- Renal aplasia
- Conn syndrome
- Essential hypertension

Description

This girl is complaining of Turner syndrome. Coarctation of the aorta is the most common cardiac complication and the probable cause of her hypertension.

CHECK ANSWER END SESSION NEXT

Session Statistics

Answered: 25 ✓ Correct: 5 × Incorrect: 20 Average Score: 20 %

Question 27 of 31

Question Difficulty: Hard

4 month old infant was admitted due to seizure for the first time. His mother reported a musty odor of his urine and sweat the started 5 days after birth.

Examination shows normally developed infant with slight hypopigmented hair and skin.

What is the most likely diagnosis?

- ○ Phenylketonuria
- ◉ Cystinuria
- ○ Homocystinuria
- ○ Galactosemia
- ○ Alport Syndrome

Description

Phenylketonuria (PKU) is an autosomal recessive condition caused due to defect in phenylalanine hydroxylase, an enzyme located on chromosome 12 which converts phenylalanine to tyrosine. Signs and symptoms include:

- Infants with PKU are normal at birth
- Seizures
- Hypopigmentation (excessively fair hair and skin)
- Musty odor of the baby's sweat and urine
- Permanent brain damage (learning difficulties and intellectual disability) occurs if PKU is untreated during the first months of life

CHECK ANSWER END SESSION NEXT

Session Statistics

Answered: 26 ✓ Correct: 6 × Incorrect: 20 Average Score: 23.08

Question 28 of 31

Question Difficulty: Hard

12 years old boy is complaining of recurrent renal stones and joints inflammation. He has intellectual disability and he has a brother of 2 brothers and 2 sisters with similar condition.

Examination shows choreiform movements of the arms and legs. Investigations show serum urea of 8.2 mmol/L (2.5-7.1 mmol/L) and knee aspiration reveals no organisms but there are numerous neutrophils containing long, needle-shaped crystals.

What is the most likely diagnosis?

- Cystinuria
- Fabry disease
- **Lesch-Nyhan syndrome**
- Phenylketonuria
- Homocystinuria

Description

Lesch-Nyhan syndrome is a rare inherited X-linked recessive disorder caused by a deficiency of the enzyme hypoxanthine-guanine phosphoribosyl transferase caused by mutations in the HPRT gene located on the X chromosome.

Signs and symptoms include:

- The HGPRT deficiency causes a build-up of uric acid in all body fluids which results in both hyperuricemia and hyperuricosuria, associated with severe gout and renal stones
- Megaloblastic anemia

Neurological signs include:

- Moderate intellectual disability
- Facial grimacing
- Involuntary writhing
- Choreiform movements of the arms and legs
- Behavioral problems

CHECK ANSWER END SESSION NEXT

Question 29 of 31

Question Difficulty: Hard

8 years old is suffering from learning and intellectual disability. He also was diagnosed to have autism. Examination reveals large protruding ears and long face. There is general muscular hypotonia with hyperextensible joints.

What is the most likely genetic diagnosis?

- 47 XYY
- Down syndrome
- Klinefelter syndrome
- Fragile X syndrome
- Acromegaly

Description

Fragile X is a trinucleotide repeat disorder; it is the most common cause of inherited mental retardation, intellectual disability and autism. Other signs and symptoms include:

- Large protruding ears
- Long face
- High-arched palate
- Flat feet
- Hyperextensible finger joints and thumbs
- Large testes in men after puberty
- Hypotonia

CHECK ANSWER END SESSION NEXT

Session Statistics

Question 30 of 31

Question Difficulty: Hard

20 years Miss X is complaining of chronic progressive skin deformities. She has a history of recurrent retinal hemorrhages and hematemesis.

Examination shows yellow skin papules in the neck and trunk. Some of them ccalesce to form plaques with reticular pattern appearance.

What is the most likely diagnosis?

- ○ Lesch-Nyhan syndrome
- ○ Pseudoxanthoma elasticum
- ○ Fragile X syndrome
- ● Hereditary hemorrhagic telangiectasia
- ○ Refsum disease

Description

Pseudoxanthoma elasticum (PXE) is a rare autosomal recessive disorder caused by mutations in the ABCC6 gene on the short arm of chromosome 16. It is manifested by:

- The lesion is yellow papules of 1-5 mm in diameter are seen in a linear or reticular pattern and may coalesce to form plaques
- Asymptomatic cutaneous elevation on the lateral aspect of the neck in the form of plucked chicken, or cobblestone appearance
- Laxity and redundant skin folds in the axilla and cubital fossa
- Eye shows angioid streaks and retinal hemorrhages in fundoscopy
- Cardiovascular manifestations as angina and hypertension
- Gastrointestinal hemorrhage with melena, frank bleeding, occult blood in the stool, or hematemesis

CHECK ANSWER END SESSION NEXT

Session Statistics

Answered: 29 ✓ Correct: 7 × Incorrect: 22 Average Score: 24.14

Question 31 of 31

Question Difficulty: Hard

Mr. X is seeking medical advice to start a family. He reported that his father has a genetic disease causing renal and ocular manifestations inherited from his mother and 2 of 3 uncles and 1 of 2 his aunts had a similar disease. He also reported all his 3 sisters have the disease and his brother is free of the disease.

What is the mode of inheritance of this disease?

- ○ Autosomal recessive inheritance
- ○ X-linked dominant inheritance
- ● Mitochondrial inheritance
- ○ X-linked recessive inheritance
- ○ Autosomal dominant inheritance

Description

The disease is Alport syndrome. It is an X-linked dominant inheritance disease. Features include:

- Males and females are both affected
- No male-to-male transmission.
- Affected males transmit the disease to 100% of their daughters.
- An affected female will transmit the disease to 50% of all offspring.

CHECK ANSWER END SESSION NEXT

Session Statistics

Answered: 30 ✓ Correct: 7 × Incorrect: 23 Average Score: 23.33 %